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CASE REPORT

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Torpedo maculopathy – a case series presentation

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ABSTRACT

Torpedo maculopathy is a lesion of the retinal pigment epithelium and/or choroid. In most cases it is unilateral, solitary and located in the temporal region of the macula. It appears hypopigmented with a hyperpigmented margin. Its shape resembles a torpedo: horizontally oval with a sharp nasal tip pointing towards the foveola and/or optic disc. The location and shape of the lesion are specific and diagnosis is clinical. The lesion is usually asymptomatic, found accidentally and does not require any treatment. However, it is associated with a risk of choroidal neovascular membrane formation, and a yearly follow-up is recommended. Torpedo maculopathy should be distinguished from congenital toxoplasmosis, congenital hypertrophy of retinal pigment epithelium, Turcot syndrome, Gardner syndrome, congenital simple hamartoma of the retina, choroidal nevus, and congenital Zika virus infection. Optical coherence tomography and fundus autofluorescence are sometimes helpful in differential diagnosis. In this paper, we aim to present four patients with torpedo maculopathy. Three of them were asymptomatic, and one experienced distortion of vision.

KEY WORDS: torpedo maculopathy, retinal lesion, fundus examination.

INTRODUCTION

Torpedo maculopathy is a congenital, nonprogressive lesion of the retinal pigment epithelium (RPE) and/or choroid [1]. It was first described by Roseman and Gass in 1992 [2]. In most cases it is asymptomatic and is found accidentally [3]. The lesion is usually unilateral and is located in the temporal region of the macula. The diagnosis of torpedo maculopathy is clinical, because its location and "torpedo-like" shape are unique. Additional imaging tests such as optical coherence tomography (OCT) and fundus autofluorescence (FAF) are useful in differential diagnosis [4]. We aim to present a case series of four patients, in whom a "torpedo-like" lesion was found in the ophthalmic investigation. All fundus images (except Figure 4) and OCT, included in this article, have been obtained with DRI OCT Trition. The FAF and fundus images in Figure 4 were captured with a Zeiss Clarus 700. FAF images in Figures 2, 6 and 8 were acquired with a mydriatic retinal camera: the Topcon TRC-50DX.

CASE REPORTS

Case 1

An 8-year-old boy was referred to the clinic for an opinion regarding a retinal lesion in his left eye that was found during a routine ophthalmic check-up. On the examination his best corrected visual acuity (BCVA) was 1.0 in both eyes. The Amsler grid test was negative in both eyes. Dilated fundus examination revealed in the left eye a "torpedo-like" hypopigmented lesion located in the temporal region of the macula with the tip pointing towards the foveola (Figure 1). We did not find any other abnormalities. OCT and FAF of the left eye were performed. OCT showed thickening of the RPE layer located in the area of the "torpedo-like" lesion (Figure 1). FAF revealed variable autofluorescence of the area corresponding to the lesion (Figure 2).

Case 2

An 11-year-old girl was referred to the clinic because of a retinal lesion of the right eye that was found on a routine check-up. She was asymptomatic. Her BCVA was 1.0 in both eyes. Fundus imaging of the right eye revealed an oval, hypopigmented lesion located in the temporal region of the macula, adjacent to the fovea (Figures 3 and 4). It is important to note that the fundus image in Figure 4. is of the same patient as in Figure 3 but was obtained with a different retinal camera (Figure 3 with DRI OCT Triton, Figure 4 with Zeiss Clarus 700). In OCT we observed outer retinal cavitation and thickening of the RPE layer temporarily to the fovea (Figure 3). FAF showed hypoautofluorescence of the lesion and a temporarily located hyperautofluorescent spot (Figure 4).

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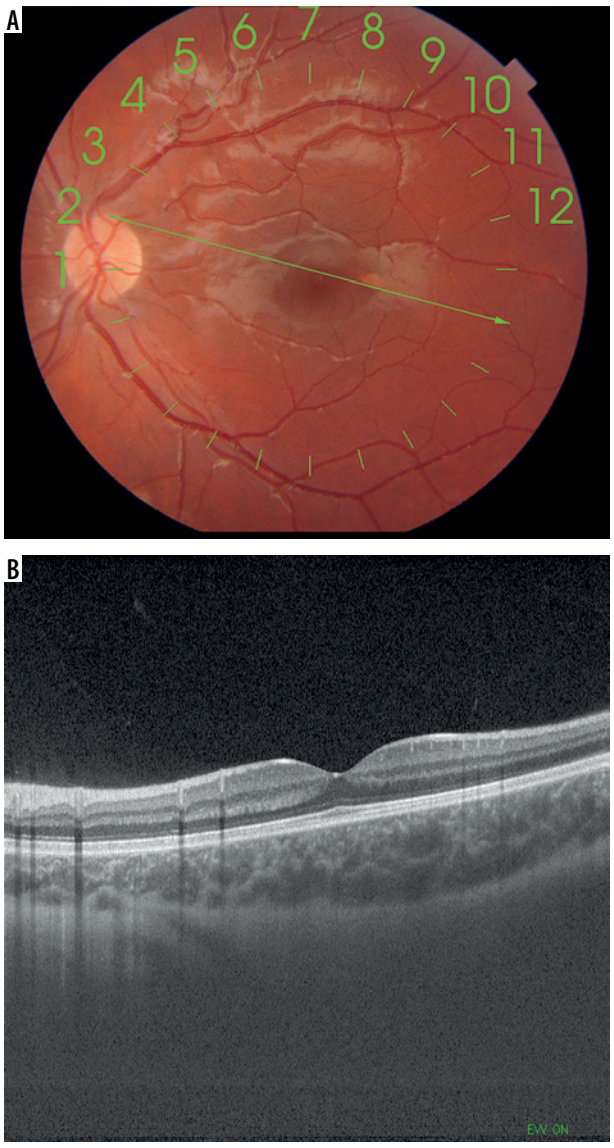

Figure 1. Case 1 – Fundus image and OCT of the left eye. Fundus photography – "torpedo-like", hypopigmented lesion with hyperpigmented margin and sharp end pointing towards foveola. OCT – thickening of RPE layer

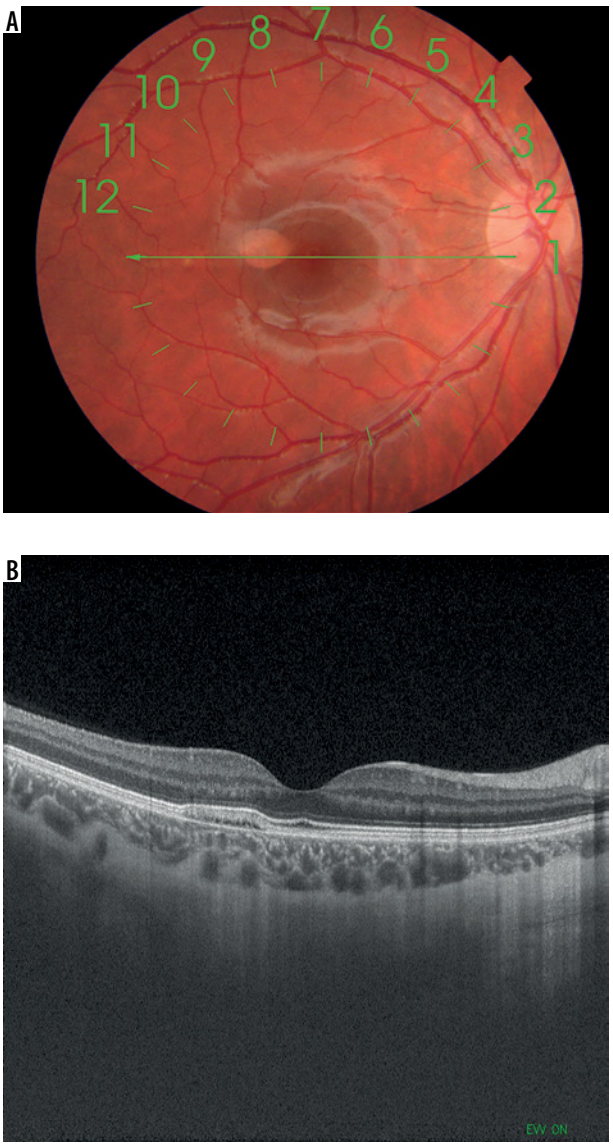

Figure 3. Case 2 – Fundus image and OCT of the right eye. Fundus photography – hypopigmented, oval lesion located in the temporal part of the macula. OCT – outer retinal cavitation and thickening of RPE layer temporarily to the fovea

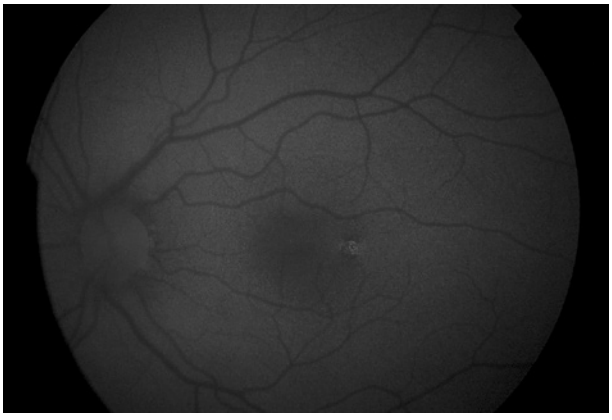

Figure 2. Case 1 – FAF of the left eye; variable autofluorescence of the area corresponding to the lesion

Case 3

A boy born at 35 weeks of gestation was first referred to the ophthalmic department for ocular consultation at the age of 17 months. He suffered from microcephaly and psychomotor retardation. He had a normal karyotype and negative tests for SMN1 gene, toxoplasmosis, Epstein-Barr virus, cytomegalovirus and common metabolic disorders. On the examination we observed, in the left eye, a hypopigmented lesion with well-defined margins located in the temporal region of the macula. At the age of 4 years his BCVA was 0.5 in the left eye and 0.6 in the right eye. The OCT showed a subtle elevation of the ellipsoid zone in the temporal region of the macula (Figure 5). FAF revealed an isoautofluorescent lesion surrounded by a hypoautofluorescent region (Figure 6).

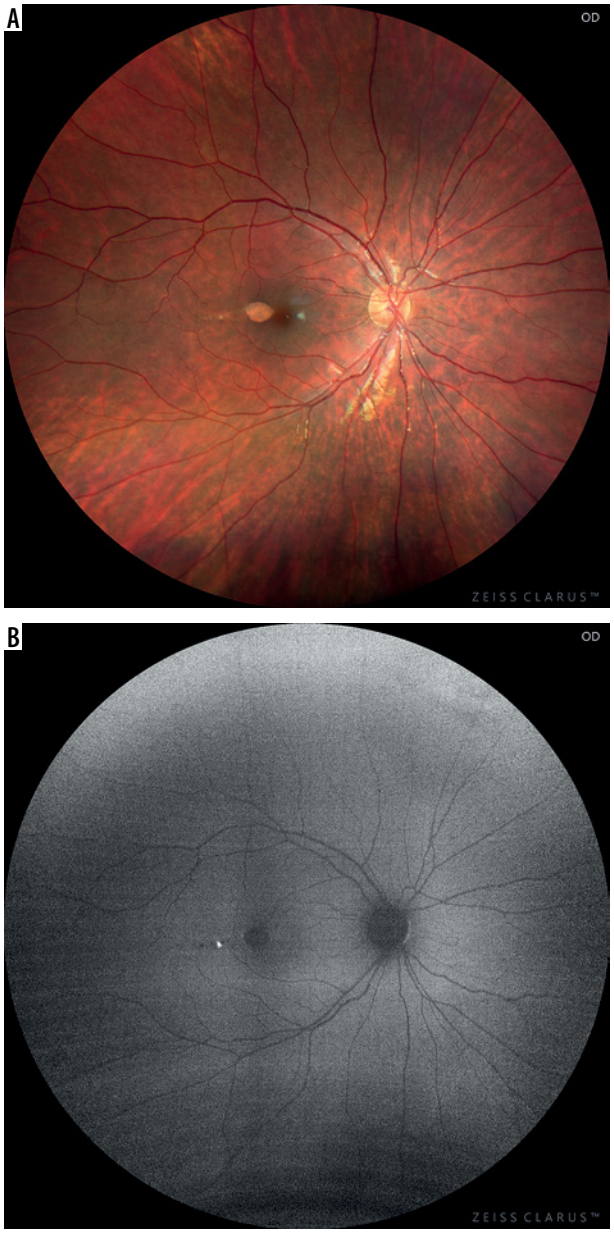

Figure 4. Case 2 – Fundus image and FAF of the right eye. Fundus image – hypopigmented, oval lesion located in the temporal part of the macula. FAF – hypoautofluorescence corresponding to the area of the lesion and hyperautofluorescent spot located temporarily

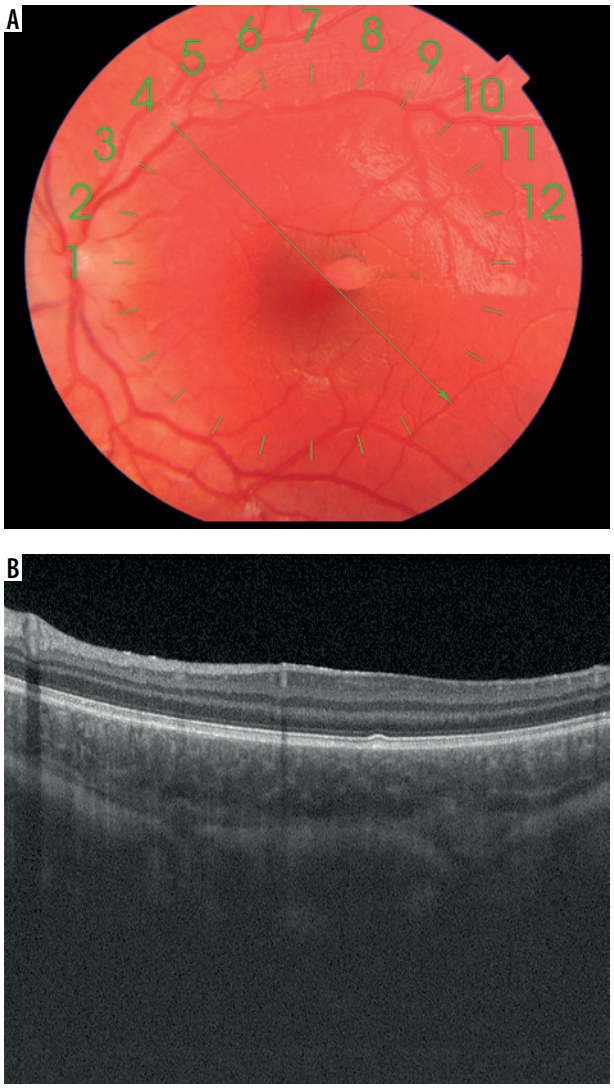

Figure 5. Case 3 – Fundus image and OCT of the left eye. Fundus image – hypopigmented, oval lesion with hyperpigmented margin and a sharp tip pointing towards the fovea, located in the temporal region of the macula. OCT – subtle elevation of the ellipsoid zone in the temporal region of the macula

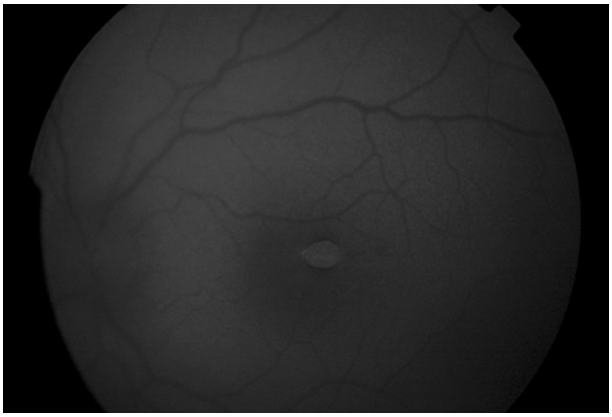

Figure 6. Case 3 – FAF of the left eye; isoautofluorescent lesion surrounded by a region of hypoautofluorescence

Case 4

A 17-year-old female patient visited our clinic for further examinations with the initial diagnosis of retinal inflammation in the left eye. Her chief complaints were "wavy vision" and headache appearing in the evenings. Her past ocular history was unremarkable. Two weeks before the visit she had a traffic accident. Her BCVA was 1.0 in both eyes. The Amsler grid test was negative for the right eye, but when looking with the left eye, she reported seeing narrowing of the lines and grids located at the left part of the chart. On the left eye fundus examination we observed a hypopigmented lesion of the RPE in the center of the macula. Its border was hyperpigmented. The test for toxoplasmosis was negative. We

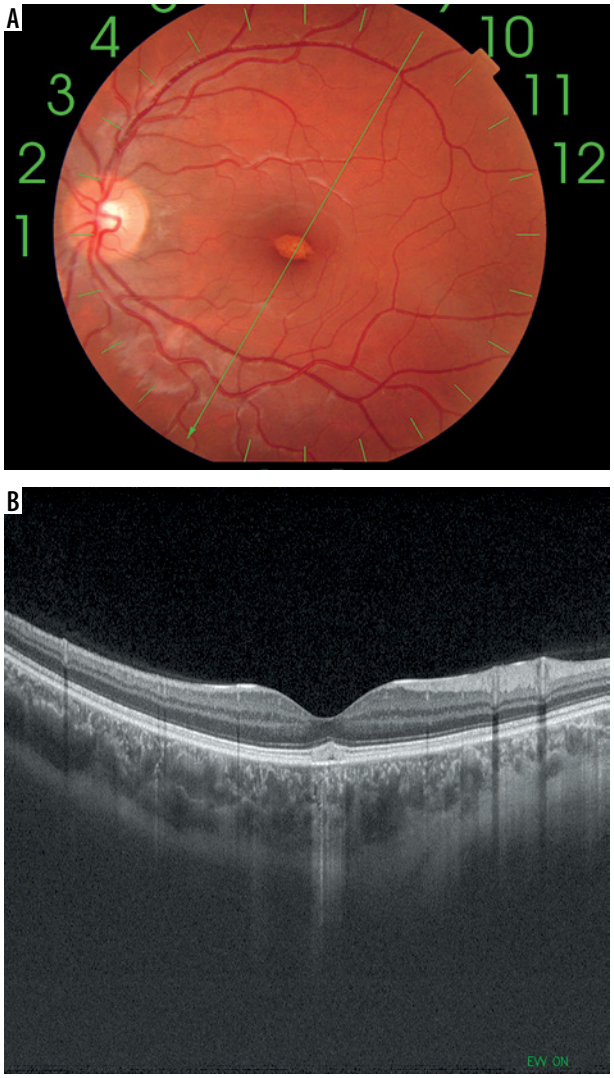

Figure 7. Case 4 – Fundus image and OCT of the left eye. Fundus photography – an oval, hypopigmented lesion with well-defined hyperpigmented margins in the center of the macula. OCT – outer retinal cavitation located in the center of the macula, underneath the foveola.

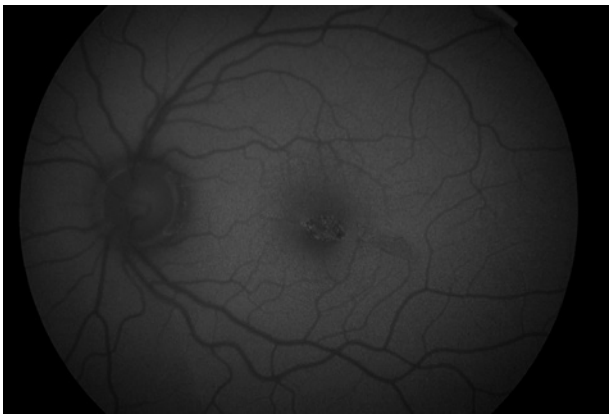

Figure 8. Case 4 – FAF of the left eye; variable autofluorescence of the lesion

performed OCT and fundus imaging (Figure 7). FAF showed variable autofluorescence of the lesion (Figure 8). Retinal inflammation was excluded and torpedo maculopathy was diagnosed. The patient has observed for the next years (2017-2020); the metamorphopsia disappeared, while OCT findings remained unchanged.

DISCUSSION

Classically, torpedo maculopathy is described as an asymptomatic, solitary, hypopigmented lesion of the RPE, located in the temporal region of the macula [4, 5]. It is horizontally oval. The nasal margin ends with a sharp tip that points into the foveola and/or optic disc [4, 6]. The temporal margin is rounded or like a hyperpigmented "frayed tail" [4]. The lesion, in shape, resembles a torpedo (an underwater missile) or a teardrop [1]. The pathogenesis of torpedo maculopathy is unclear. There are some theories that have been proposed: congenital defects in the development of the nerve fiber layer, abnormal development of choroidal or ciliary vasculature or persistent defect in the development of the RPE in the fetal temporal bulge [4, 6–8]. There have been noted correlations that support a congenital retinal nerve fiber layer-driven etiology, namely: most of the lesions points toward the optic nerve; lesion horizontal angularity is consistent with distribution of the horizontal raphe of the retinal nerve fiber layer; and the nasal margin localizes at the junction of the superior arcuate, inferior arcuate and papillomacular bundles of the retinal fiber layer. Moreover, it has been observed, based on retinal laser ellipsometry, that the nasal well-shaped margin of the lesion is associated with relatively low density of nerve fiber layer. This aspect may explain the consistent localization of torpedo maculopathy [6]. Analysis of the literature proves that the manifestation of torpedo maculopathy is diverse. There have been reported cases in which two "torpedo-like" lesions were present in one eye or the main lesion was accompanied by satellite lesions [3, 9]. Location of the lesion also varies. It has been found outside the macula (in the temporal region of the retina, inferior to the macula, inferonasal to the optic disc) or in its center [3, 10–12]. However, some characteristics, in most cases, remained unchanged, i.e.: an oval, torpedo-like shape and a sharp tip pointing towards the foveola [4, 10]. Standard automated perimetry may reveal the presence of a scotoma corresponding to the location of the lesion, but in some cases, a scotoma can only be detected in microperimetry [13]. Cases of torpedo maculopathy that coexisted with retinoblastoma have also been described [5]. Although the diagnosis of torpedo maculopathy is made clinically, OCT and FAF may be helpful to support the diagnosis and exclude other disorders. Based on OCT images, four types of lesions have been identified [1, 14, 15]. In 2015 Wong *et al.* classified lesions, in patients with torpedo maculopathy, into two types: type 1 – lesion with attenuation of outer retinal structures but without presence of outer retinal cavitation; and type 2 – lesion with both attenuation and cavitations of outer retinal structures [14]. In 2018 Tripathy *et al.* introduced a new, third type: degeneration of the outer retina accompanied with excavation of the inner retina and choroid [15]. The fourth type was described in 2020 by Light *et al.* and it represents a lesion with a preserved ellipsoid zone, lack of subretinal fluid, but

with inner choroidal excavation [1]. FAF usually shows uniform or variable hypoautofluorescence at the area of the lesion and small regions of hyperautofluorescence at its margins [12, 14]. Usually torpedo maculopathy is asymptomatic, non-progressive and does not require any treatment [4]. However, the risk of choroidal neovascular membrane formation is an important consideration for long-term follow-up [5]. In all of our patients, we have identified unilateral, torpedo-like lesions. In three cases the lesion was located in the left eye. Three of them did not involve a fovea and were asymptomatic. However, in case 4 the lesion was located in the fovea, which resulted in distorted vision. According to the OCT classification, we can classify our patients into type 1 (cases 1 and 3) and type 2 (cases 2 and 4). None of our patients had type 3 or 4 lesions. FAF has been performed in all cases (Figures 2, 4, 6 and 8). In cases 1 and 4 it revealed variable autofluorescence of the lesion. In case 2 the lesion was hypoautofluorescent and there was a hyperautofluorescent spot located temporarily to the macula. In case 3, an isoautofluorescent lesion was surrounded by a region of hypoautofluorescence. Patients have not been provided with any treatment. However, we recommended a yearly follow-up. Torpedo maculopathy should be distinguished from other retinal disorders, congenital and acquired. The differential diagnosis includes congenital toxoplasmosis, congenital hypertrophy of RPE, Turcot syndrome, Gardner syndrome, congenital simple hamartoma of the retina, choroidal nevus, and congenital Zika virus infection [6, 13] the underlying etiology of torpedo maculopathy has remained elusive. In this literature review, we provide new evidence to better support, reject and unify claims regarding cause, diagnosis, and proper clinical management of this disease. We reviewed 44 case reports and case series, which included 77 patients (after exclusions. Congenital toxoplasmosis manifests as retinochoroidal lesions. Active lesions appear as whitish foci, which after healing turn into scars [16]. It can be challenging to distinguish between torpedo maculopathy and toxoplasmosis on fundus examination. To exclude or confirm ocular toxoplasmosis one should perform serologic tests [17]. Congenital hypertrophy of the RPE (CHRPE) is a pigmented, flat, round or oval lesion that can be found at the mid-periphery or at the posterior pole. There are regions of RPE loss in the lesion area, which appear as depigmented lacunae. In OCT the region that underlies the lesion appears as loss of photoreceptors and outer retinal layers [18]. Fundus autofluorescence shows hypoautofluorescence with lacunae being isoautofluorescent or hypoautofluorescent [19]. Fluorescein angiography shows choroidal masking. Also, CHRPE can be a specific and sensitive marker of adenomatous polyposis of the colon (APC) or familial adenomatous polyposis (FAP) [18]. The lesions found in patients with FAP or APC resemble CHRPE but they are multifocal (more than three), bilateral, mixed (pigmented and depigmented), variable in shape (ovoid, pisciform, irregular, surrounded by depigmented halo), size (from dot-like to multiple disc diameters) and location (midperiphery, near the optic disc or macula) [18, 20-22]. Traboulsi and colleagues termed these lesions associated with polyposis syndrome as pigmented ocular fundus lesions (POFLs) [21, 23]. FAP may be present in Gardner and Turcot syndromes. Turcot syndrome is characterized by the coexistence of a primary central nervous system tumor (usually medulloblastoma or glioblastoma multiforme), numerous adenomatous colorectal polyps (APC) and colonic adenocarcinoma [24]. Gardner syndrome is an autosomal dominant subtype of FAP. It is characterized by adenomatous intestinal polyps, multiple osteomas in the skull, maxillae, mandible and cutaneous and subcutaneous masses [22, 25]. A congenital simple hamartoma of the retina (CSHRPE) is a rare, black, benign tumor; it is located in a macula adjacent to the fovea. It penetrates through all layers of the retina, may protrude into the vitreous and is usually accompanied by a feeder arteriole and venule [18, 20]. Sometimes, the lesion is complicated with retinal traction or intraretinal hemorrhages, which decreases visual acuity. OCT shows elevation of the inner retina and marked optical shadowing at the level of the outer retina and choroid, with normal retina adjacent to the lesion [18]. Choroidal nevus is round or oval and its color is yellowish to brown. In a chronic lesion OCT shows the adjacent RPE to be atrophic, hypertrophic or metaplastic and to be accompanied with drusen [20]. Congenital Zika virus is associated with microcephaly and may affect the posterior segment of the eye. The ocular manifestations that have been reported include macular chorioretinal atrophy, RPE mottling, optic nerve hypoplasia, atrophy, pallor and increased cup-to-disc ratio. Most of the findings occurred bilaterally [26-29]. None of our patients suffered from any systemic or genetic disease. All of them had a hypopigmented, unilateral lesion. In patients 3 and 4 we excluded toxoplasmosis infection. In patients 1 and 2 the temporal location and torpedo-like shape of the lesion enabled us to make a clinical diagnosis.

CONCLUSIONS

We present four patients diagnosed with torpedo maculopathy. In all patients the lesion was unilateral. All patients had attenuation of outer retinal layers on OCT. In most cases the diagnosis of torpedo maculopathy can be made clinically. Additional imaging tests such as OCT and FAF are sometimes helpful in differential diagnosis. In the long-term follow-up it is important to consider the risk of choroidal neovascular membrane formation.

DISCLOSURE

The authors declare no conflicts of interest.

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